



SH3TC2 gene

SH3 domain and tetratricopeptide repeats 2

Normal Function

The *SH3TC2* gene is active in the nervous system and provides instructions for making a protein whose function is unknown. Based on its structure, the SH3TC2 protein probably interacts with other proteins and may assist in assembling proteins into a group or complex.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

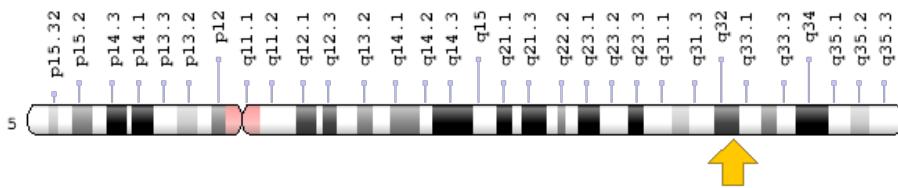
Researchers have identified at least 19 *SH3TC2* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 4C. Charcot-Marie-Tooth disease is a group of progressive disorders that affect the peripheral nerves. Peripheral nerves connect the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch, pain, heat, and sound.

Most of the *SH3TC2* gene mutations that cause Charcot-Marie-Tooth disease disrupt production of the SH3TC2 protein, resulting in an abnormally short or unstable version of this protein. Some mutations change one of the building blocks (amino acids) used to make the SH3TC2 protein, which alters the protein's structure. It is unclear how *SH3TC2* gene mutations cause the signs and symptoms of type 4C Charcot-Marie-Tooth disease.

Chromosomal Location

Cytogenetic Location: 5q32, which is the long (q) arm of chromosome 5 at position 32

Molecular Location: base pairs 148,982,150 to 149,063,174 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT4C
- CMTND
- FLJ13605
- KIAA1985
- S3TC2_HUMAN

Additional Information & Resources

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 4C
<https://www.ncbi.nlm.nih.gov/books/NBK1340>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SH3TC2%5BTIAB%5D%29+OR+%28SH3/TPR%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SH3 DOMAIN AND TETRATRICCOPEPTIDE REPEAT DOMAIN 2
<http://omim.org/entry/608206>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SH3TC2%5Bgene%5D>
- HGNC Gene Family: Tetra-tricopeptide repeat domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/769>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=29427
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=29>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/79628>
- UniProt
<http://www.uniprot.org/uniprot/Q8TF17>

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